CLAIMS

1. A method of detection of human Spinocerebellar ataxia 2 gene variants and the said method comprises:
   a) designing and synthesizing oligonucleotide primers for PCR amplification of CAG repeat containing region of exon 1 of human SCA2 gene,
   b) amplifying genomic DNA of SCA2 patients and normal control individuals using the said primers of step (a),
   c) sequencing the amplified PCR product and identifying the sequence variations computationally by comparing it with the already existing sequence of human SCA2 gene,
   d) screening normal control individuals and SCA2 patients for novel single nucleotide polymorphisms using allele specific oligonucleotide probes,
   e) computing the frequencies of CC and GT haplotypes in normals and SCA2 patients,
   f) establishing the association of the CC and GT haplotype with the SCA2 disease based on their frequency distribution in normals and SCA2 patients, and
   g) predicting the risk or susceptibility to the SCA2 disease based on the haplotype present at the polymorphic sites in the individual tested, GT haplotype being at low risk and CC haplotype at high risk to the disease.

2. A method as claimed in claim 1 wherein, the primers suitable for amplification of the SCA2 gene region containing one or more polymorphic sites, are selected from the group.
   a) CTC CGC CTC AGA CTG TTT TGG TAG 3' (as listed in SEQ ID NO: 1).
   b) GTG GCC GAG GAC GAG GAG AC 3' (as listed in SEQ ID NO: 2) and compliments thereof.
3. A method as claimed in claim 1 wherein, the allele specific primers suitable for detection of the allelic variants of SCA2 gene are selected from:
   a) 5' CTC GGC GGG CCT CCC CGC CCC TTC GTC GTC C 3' (as listed in SEQ ID NO: 3);
   b) 5' CTC GGC GGG CCT CCC CGC CCC TTC GTC GTC G 3' (as listed in SEQ ID NO: 4);

c) 5' CCT CCC CGC CCC TTC GTC GTC 3' (as listed in SEQ ID NO: 5);

d) 5' CGC CAA CCC GCG CCT CCC CGC TCG GCG CCC GC 3' (as listed in SEQ ID NO: 6);

e) 5' CGC CAA CCC GCG CCT CCC CGC TCG GCG CCC GT 3' (as listed in SEQ ID NO: 7); and

f) 5' GCG CCT CCC CGC TCG GCG CCC G 3' (as listed in SEQ ID NO: 8) and compliments thereof.

4. A method as claimed in claim 1 wherein, the allele specific probes useful for detection of SCA2 gene variants, wherein the polymorphic site occupies a central position of the probe are selected from:

a) 5' CCC CTT CGT CGT CCT CCT TCT CCC CCT 3' (as listed in SEQ ID NO: 9);

b) 5' CCC CTT CGT CGT CGT CCT TCT CCC CCT 3' (as listed in SEQ ID NO: 10);

c) 5' CGC TCG GCG CCC GCG CGT CCC CGC CGC 3' (as listed in SEQ ID NO: 11); and

d) 5' CGC TCG GCG CCC GTG CGT CCC CGC CGC 3' (as listed in SEQ ID NO: 12) are compliments thereof.

5. A method as claimed in claim 1 wherein, the length of the oligonucleotide primers and probes of claims 2, 3 and 4 is in the range of 5 to 100 bases.

6. A diagnostic kit for the detection of SNP haplotypes (CC/GT) comprising suitable primers and probes are selected from the group consisting of sequences given under SEQ ID NO: 1 to 12.

7. Primers suitable for amplification of SCA2 gene region containing one or more polymorphic sites, said primers selected from the group comprising :

a) CTC CGC CTC AGA CTG TTT TGG TAG 3' (as listed in SEQ ID NO: 1); and

b) GTG GCC GAG GAC GAG GAG AC 3' (as listed in SEQ ID NO: 2) and compliments thereof.

8. Allele specific primers suitable for detection of allelic variants of SCA2 gene, selected from the group comprising:

a) 5' CTC GGC GGG CCT CCC CGC CCC TTC GTC GTC C 3' (as listed in SEQ ID NO: 3);

b) 5' CTC GGC GGG CCT CCC CGC CCC TTC GTC GTC G 3' (as listed in SEQ ID NO: 4);

c) 5' CCT CCC CGC CCC TTC GTC GTC 3' (as listed in SEQ ID NO: 5);

d) 5' CGC CAA CCC GCG CCT CCC CGC TCG GCG CCC GC 3' (as listed in SEQ ID NO: 6);

e) 5' CGC CAA CCC GCG CCT CCC CGC TCG GCG CCC GT 3' (as listed in SEQ ID NO: 7); and

f) 5' GCG CCT CCC CGC TCG GCG CCC G 3' (as listed in SEQ ID NO: 8) and compliments thereof.

9. Allele specific probes useful for detection of SCA2 gene variants wherein the polymorphic site occupies a central position of the probe, said allele specific probes selected from the group comprising:

a) 5' CCC CTT CGT CGT CCT CCT TCT CCC CCT 3' (as listed in SEQ ID NO: 9);

b) 5' CCC CTT CGT CGT CGT CCT TCT CCC CCT 3' (as listed in SEQ ID NO: 10);

c) 5' CGC TCG GCG CCC GCG CGT CCC CGC CGC 3' (as listed in SEQ ID NO: 11); and

d) 5' CGC TCG GCG CCC GTG CGT CCC CGC CGC 3' (as listed in SEQ ID NO: 12) are compliments thereof.